INSTITUTE OF IMMUNOLOGY AND GENETICS

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Requesting physician:	**Patient information:**

Request Form cardiovascular diseases and RASopathy

Cause of examination: ☐ diagnostic ☐ predictive for familial mutation _____

Clinical information:

Family history:

(Check all that apply)

☐ Patient clinically affected

☐ No previous molecular genetic examinations existent

☐ The following previous molecular genetic examinations have been done:

_____

Date / Time of sampling:	**Signature:**

Specimen requirements and logistics:

- **Patient Consent Form for Genetic Testing according to the German Law and Request Form**
- **Billing information (Insurance-, Institutional- or Selfpay-Billing)**
- **5 ml EDTA blood collection tube labeled with patient name and date of birth**

Patient name: _____ **DOB:** _____

☐ **Arrhythmogenic right ventricular cardiomyopathy (ARVC)**
Step 1: *CDH2, DES, DSC2, DSG2, DSP, FLNC, JUP, LMNA, PKP2, PLN, TMEM43*
Step 2: *ANK2, PRDM16, RYR2*

☐ **Dilated Cardiomyopathy (DCM)**
Step 1: *ACTN2, ANKRD1, BAG3, DES, DMD, LDB3, LMNA, MYBPC3, MYH7, PRDM16, RBM20, SCN5A, SGCD, TCAP, TMPO, TNNI3, TNNT2, TPM1*
Step 2: *ACTC1, CSRP3, DSG2, DSP, EMD, EYA4, FHL1, HFE, MYH6, NXN, PLN, PSEN1, PSEN2, TAZ, TTN, VCL*

☐ **Hypertrophic Cardiomyopathy (HCM)**
Step 1: *ACTC1, MYBC3, MYH7, MYL2, PLN, TNNI3, TNNT2, TPM1*
Step 2: *ACTN2, ALPK3, ANKRD1, CALR3, CSRP3, FLNC, JPH2, KLF10, MYH6, MYLK2, MYOM1, MYPN, NXN, OBSC, PDLIM3, RYR2, TCAP, TNNC1, TRIM63, TTN, VCL*
Step 3: *BRAF, GAA, GLA, HRAS, LAMP2, LZTR1, MAP2K1, NRAS, NPC1, PAHX, PRKAG2, PTPN11, RAF1, RIT1, SOS1, TTR*

☐ **Left ventricular non-compaction Cardiomyopathy (LVNC)**
Step 1: *ACTC1, ACTN2, CASQ2, DSP, DTNA, LDB3, LMNA, MIB1, MYBPC3, MYH7, PRDM16, RYR2, SCN5A, TAZ, TNNT2, TPM1*
Step 2: *ABCC9, DMPK, HCN4, PKP2, PLEKHM2*

☐ **Restrictive Cardiomyopathy (RCM)**
Step 1: *ACTC1, BMP5, BMP7, CRYAB, DES, DNAJB6, FHL1, FLNC, HOGA1, IDS, IDUA, LBD3, LMNA, MYBPC3, MYH7, TNNI3, TNNT2*
Step 2: *ABCC6, AGXT, APOA1, GBA, GLA, GRHPR, HAMP, HFE, HFE2, HJV, MYOT, MYPN, NCP1, NCP2, PNPLA3, SLC40A1, SMPD1, TAZ, TNNC1, TNNI3, TNNT2, TTR, WRN*

☐ **Brugada-Syndrome**
Step 1: *ABCC9, CACNA1C, CACNA2D1, CACNB2, GPD1L, HCN4, KCNE3, KCNE5, SCN1B, SCN5A, SCN10A, TRPM4*
Step 2: *FGF12, KCND2, KCND3, KCNH2, KCNJ8, PKP2, RANGRF, SCN2B, SCN3B, SEMA3A, SLMAP*

☐ **Catecholaminergic polymorphic ventricular tachycardia (CPVT)**
CALM1, CASQ2, RYR2, TRDN

☐ **Long-QT-Syndrome (LQTS)**
Step 1: *KCNE1, KCNH2, KCNQ1, SCN5A*
Step 2: *AKAP9, ANK2, CACNA1C, CALM1, CALM2, CAV3, KCNE2, KCNJ2, KCNJ5, SCN4B, SNTA1*

☐ **Short-QT-Syndrome (SQTS)**
Step 1: *KCNH2, KCNJ2, KCNQ1, SLC4A3*
Step 2: *CACNA1C, CACNB2, SCN5A*

☐ **Isolated congenital heart defect**
Step 1: *ACTC1, ACTA2, CITED2, COL1A1, CREDL1, ELN, FBN2, FGFR2, FLNA, GATA4, GATA6, GDF1, GJA1, JAG1, MYH11, MYH6, MYH7, NKX2-5, NOTCH1, NR2F2, TBX5, ZIC3*
Step 2: *ABCC9, ACVR2B, ADAMTS19, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CFAP53, CFC1, DNAAF3, DYX1C1, LEFTY2, MMP21, NODAL, NOTCH2, SALL4, SOS1,*

☐ **Syndromic congenital heart defect**
Step 1: *ARID1A, ASXL1, CDK13, CHD4, CHD7, CREBBP, CRELD1, DHCR7, EHMT1, ELN, EP300, EVC, EVC2, KANSL1, KDM6A, KMT2A, KMT2D, MED13L, NSD1, PRKD1, RAI1, SLC2A10, SOX2, SOX9, TAB2, TBX1, TBX20, TBX5*
Step 2: *ABCC9, ACTB, ADNP, AMER1, BBS2, BBS6, BCOR, BRAF, BRAF1, C14orf104, C19orf51, CBL, CCBE1, CCDC103, CCDC39, CCDC40, CD96, CHST14, COL1A1, COL3A1, DNAH11, DNH5, DNAI1, DNAI2, DNAL1, DYX1C1, EFTUD2, FBN2, FGFR2, FLNA, FLNB, FOXC1, FOXC2, FOXF1, GLI3, GPC3, HCCS, HDAC4, HRAS, IFT122, IFT140, INVS, IRX5, JAG1, KRAS, LRRC50, LTBP3, MAP2K1, MAP2K2, MED12, MEGF8, MGP, MID1, MKKS, MKS1, NEK1, NEK8, NF1, NFATC1, NIPBL, NME8, NOTCH1, NOTCH2, NPHP3, NRAS, PEX1, PEX10, PEX2, PEX26, PEX5, PKD1, PTPN11, RAB23, RBM8A, RIT1, SALL1, SALL4, SHOC2, SH3PXD2B, SKI, SMAD3, SMAD4, SMARCA2, SMARC4, SMARCB1, SMARCE1, SMC1A, SOS1, STRA6, TBX3, TFAP2B, TGFBR1, TGFBR2, UBR1, ZEB2, ZIC3*

☐ **RASopathy**

- ☐ Noonan-Syndrome: *CBL, KRAS, LZTR1, NRAS, PTPN11, RAF1, RASA2, RIT1, RRAS, SHOC2, SOS1, SOS2 MSH2, MSH6*
- ☐ CFC-Syndrome: *BRAF, KRAS, MAP2K1, MAP2K2*
- ☐ Costello-Syndrome: *HRAS*
- ☐ LEOPARD-Syndrome: *BRAF, MAP2K1, PTPN11, RAF1*
- ☐ Legius-Syndrome: *SPRED1*